ARMS2 gene

age-related maculopathy susceptibility 2

Normal Function

The *ARMS2* gene provides instructions for making a protein whose function is unknown. Studies suggest that the ARMS2 protein is found primarily in the placenta and in the specialized light-sensing tissue in the back of the eye (the retina). However, it is unclear what role, if any, the protein plays in early development or normal vision.

Health Conditions Related to Genetic Changes

age-related macular degeneration

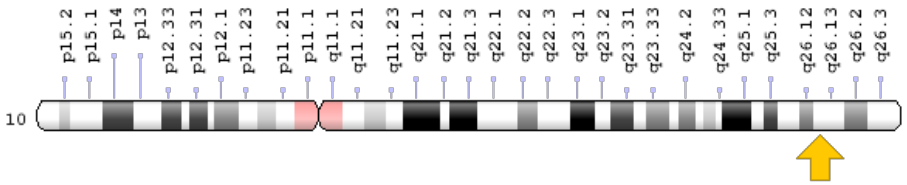
The *ARMS2* gene is located on the long (q) arm of chromosome 10 in a region known as 10q26. This region has been strongly associated with the risk of developing age-related macular degeneration, a common cause of vision loss in older adults. Researchers have identified several variations (polymorphisms) in and near the *ARMS2* gene that may explain the association between the 10q26 region and age-related macular degeneration. The best-studied of these variations, known as rs10490924, alters a single protein building block (amino acid) in the ARMS2 protein. Another common variation, a complex change that deletes a segment of the *ARMS2* gene and inserts new genetic material, may also contribute to disease risk.

It is unclear how polymorphisms in the *ARMS2* gene might be related to age-related macular degeneration. In the 10q26 region, the *ARMS2* gene is located next to a gene called *HTRA1*; changes in this gene have also been studied as a risk factor for the disease. Because the two genes are so close together, it is difficult to tell whether changes in one gene or the other, or possibly changes in both genes, account for the increased disease risk. Age-related macular degeneration is a complex condition that likely results from a combination of multiple genetic and environmental factors.

Chromosomal Location

Cytogenetic Location: 10q26.13, which is the long (q) arm of chromosome 10 at position 26.13

Molecular Location: base pairs 122,454,663 to 122,457,352 on chromosome 10 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- age-related maculopathy susceptibility protein 2
- ARMD8
- ARMS2_HUMAN
- LOC387715

Additional Information & Resources

Educational Resources

- Neuroscience (second edition, 2001): Macular Degeneration https://www.ncbi.nlm.nih.gov/books/NBK10850/box/A754/
- Webvision: The Organization of the Retina and Visual System (2008): Molecular genetics of AMD https://www.ncbi.nlm.nih.gov/books/NBK27323/#maculardegen.Molecular_genetics_of_AMD

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ARMS2%5BTIAB%5D%29+OR+%28LOC387715%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- ARMS2 GENE
  http://omim.org/entry/611313

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ARMS2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ARMS2%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=32685
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/387715
- UniProt
  http://www.uniprot.org/uniprot/P0C7Q2

Sources for This Summary

- Friedrich U, Myers CA, Fritsche LG, Milenkovich A, Wolf A, Corbo JC, Weber BH. Risk- and non-risk-associated variants at the 10q26 AMD locus influence ARMS2 mRNA expression but exclude pathogenic effects due to protein deficiency. Hum Mol Genet. 2011 Apr 1;20(7):1387-99. doi: 10.1093/hmg/ddr020. Epub 2011 Jan 20.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21252205
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3049360/
- Fritsche LG, Loenhardt T, Janssen A, Fisher SA, Rivera A, Keilhauer CN, Weber BH. Age-related macular degeneration is associated with an unstable ARMS2 (LOC387715) mRNA. Nat Genet. 2008 Jul;40(7):892-6. doi: 10.1038/ng.170. Epub 2008 May 30.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18511946
- Kanda A, Chen W, Othman M, Branham KE, Brooks M, Khanna R, He S, Lyons R, Abecasis GR, Swaroop A. A variant of mitochondrial protein LOC387715/ARMS2, not HTRA1, is strongly associated with age-related macular degeneration. Proc Natl Acad Sci U S A. 2007 Oct 9;104(41):16227-32. Epub 2007 Sep 20.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17884985
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1987388/
- Kanda A, Stambolian D, Chen W, Curcio CA, Abecasis GR, Swaroop A. Age-related macular degeneration-associated variants at chromosome 10q26 do not significantly alter ARMS2 and HTRA1 transcript levels in the human retina. Mol Vis. 2010 Jul 15;16:1317-23.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20664794
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2905635/

- Rivera A, Fisher SA, Fritsche LG, Keilhauer CN, Lichtner P, Meitinger T, Weber BH. Hypothetical LOC387715 is a second major susceptibility gene for age-related macular degeneration, contributing independently of complement factor H to disease risk. Hum Mol Genet. 2005 Nov 1; 14(21):3227-36. Epub 2005 Sep 20.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16174643
- Tong Y, Liao J, Zhang Y, Zhou J, Zhang H, Mao M. LOC387715/HTRA1 gene polymorphisms and susceptibility to age-related macular degeneration: A HuGE review and meta-analysis. Mol Vis. 2010 Oct 5;16:1958-81. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21031019
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2956667/
- Yang Z, Tong Z, Chen Y, Zeng J, Lu F, Sun X, Zhao C, Wang K, Davey L, Chen H, London N, Muramatsu D, Salasar F, Carmona R, Kasuga D, Wang X, Bedell M, Dixie M, Zhao P, Yang R, Gibbs D, Liu X, Li Y, Li C, Li Y, Campochiaro B, Constantine R, Zack DJ, Campochiaro P, Fu Y, Li DY, Katsanis N, Zhang K. Genetic and functional dissection of HTRA1 and LOC387715 in age-related macular degeneration. PLoS Genet. 2010 Feb 5;6(2):e1000836. doi: 10.1371/journal.pgen.1000836.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20140183
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2816682/

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